American Journal of Medical Genetics 92:296–297 (2000)

Letter to the Editor

De Novo Partial Duplication of Long Arm of Chromosome 13: dup(13)(q12→q14)

To the Editor:

Partial chromosome duplications usually occur de novo, in most cases during meiosis. Their cytogenetic and molecular analysis assists in mapping certain phenotypes to specific chromosomal regions. The trisomy of region (13q14→qter) results in somewhat similar, but much less severe clinical symptoms as compared with the full trisomy of chromosome 13 [Hornstein et al., 1981; Koske-Westphal et al., 1978; Schwartz et al., 1991]. The critical region for the trisomy 13 syndrome has been suggested to be at region 13q14-q32 [Helali et al., 1996].

We report on a 7-year-old girl who was referred for short stature and mental retardation. The family history was unremarkable. The patient was born at term with normal growth parameters. Coarctation of the aorta was corrected at age 2 years. She had no subsequent significant health problems. Early developmental delay was considered to be secondary to the cardiac surgery. She started to walk at the age 2 years, and to talk at age 4 years. At examination her height and weight were below the fifth centile, head circumference at the tenth centile. She had relative macrocephaly, high forehead, telecanthus, epicanthus, hypoplastic midface, coarse hair, and ataxic and mildly spastic gait.

Routine and high resolution G-banded chromosome studies were performed demonstrating extra chromosomal material at the subcentromeric region of the long arm of chromosome 13. C-banding showed a normal centromeric region. High-resolution studies showed no chromosomal abnormality in the patient's parents. To confirm the partial duplication of chromosome 13, fluorescence in situ hybridization (FISH) studies were done with the chromosome 13 library (Oncor). This confirmed that the extra chromosomal material was of 13 in origin. Studies with the chromosome 13 alpha-satellite specific FISH probe (D13Z3, Oncor) showed no alteration. Since the cytogenetic results suggested a duplication in the 13q12-14 region, further hybridization was performed with the retinoblastoma susceptibility gene (13q14) specific probe (LSI13,Oncor), demonstrating a duplication of this region in our patient (Fig. 1).

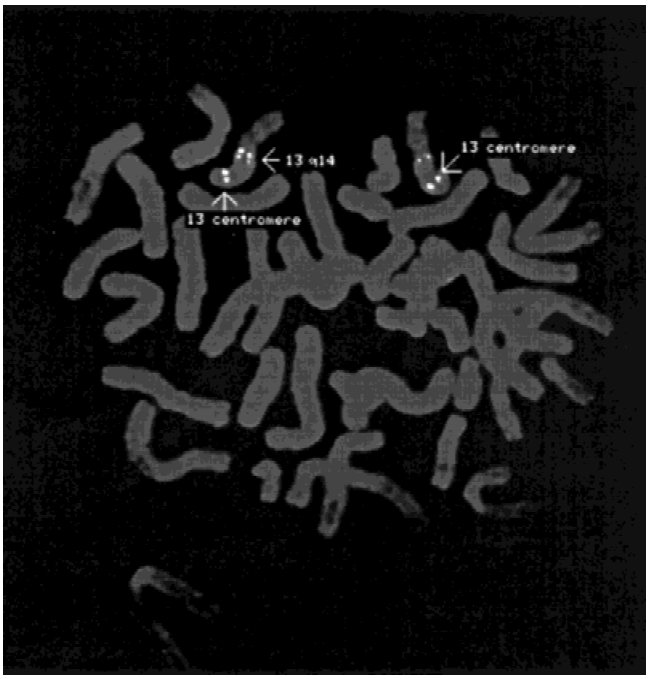

Fig. 1. FISH study with the 13q14 region specific fluorescent cosmid probe and a control centromere probe (Oncor) demonstrating a duplication of the 13q14 region.

Our patient had none of the characteristics of trisomy 13, thus we suggest that the critical region for the trisomy 13 syndrome is narrowed down to 13q21-q32. As additional benefit for our patient, this finding warrants continuous clinical follow up, since an increased susceptibility for retinoblastoma and osteosarcoma may be predicted in various alterations (mostly deletions) of the 13q14 chromosomal region [Zhu et al., 1989].

Grant sponsor: OTKA; Grant number: T 29888/1999.

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Received 29 September 1999; Accepted 15 February 2000

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